Cohen syndrome

Cohen syndrome is an inherited disorder that affects many parts of the body and is characterized by developmental delay, intellectual disability, small head size (microcephaly), and weak muscle tone (hypotonia). Other features include progressive nearsightedness (myopia), degeneration of the light-sensitive tissue at the back of the eye (retinal dystrophy), an unusually large range of joint movement (hypermobility), and distinctive facial features. Characteristic facial features include thick hair and eyebrows, long eyelashes, unusually-shaped eyes (down-slanting and wave-shaped), a bulbous nasal tip, a smooth or shortened area between the nose and the upper lip (philtrum), and prominent upper central teeth. The combination of the last two facial features results in an open-mouth appearance.

The features of Cohen syndrome vary widely among affected individuals. Additional signs and symptoms in some individuals with this disorder include low levels of white blood cells (neutropenia), overly friendly behavior, and obesity that develops in late childhood or adolescence. When obesity is present, it typically develops around the torso, with the arms and legs remaining slender. Individuals with Cohen syndrome may also have narrow hands and feet, and slender fingers.

Frequency

The exact incidence of Cohen syndrome is unknown. It has been diagnosed in fewer than 1,000 people worldwide. More cases are likely undiagnosed.

Genetic Changes

Mutations in the *VPS13B* gene (frequently called the *COH1* gene) cause Cohen syndrome. The function of the protein produced from the *VPS13B* gene is unknown; however, researchers suggest it may be involved in sorting and transporting proteins inside the cell. Most mutations in the *VPS13B* gene are believed to prevent cells from producing a functional VPS13B protein. It is unclear how loss of functional VPS13B protein leads to the signs and symptoms of Cohen syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Hypotonia, obesity, and prominent incisors
- Norio syndrome
- obesity-hypotonia syndrome
- Pepper syndrome
- prominent incisors-obesity-hypotonia syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Cohen syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265223/

Other Diagnosis and Management Resources

- GeneReview: Cohen Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1482
- MedlinePlus Encyclopedia: Hypotonia
  https://medlineplus.gov/ency/article/003298.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Hypotonia
  https://medlineplus.gov/ency/article/003298.htm
- Health Topic: Craniofacial Abnormalities
  https://medlineplus.gov/craniofacialabnormalities.html
- Health Topic: Obesity
  https://medlineplus.gov/obesity.html

Genetic and Rare Diseases Information Center

- Cohen syndrome
  https://rarediseases.info.nih.gov/diseases/6126/cohen-syndrome

Educational Resources

- Disease InfoSearch: Cohen Syndrome
  http://www.diseaseinfosearch.org/Cohen+Syndrome/1715
- MalaCards: cohen syndrome
  http://www.malacards.org/card/cohen_syndrome
- Orphanet: Cohen syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=193

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/cohen-syndrome/

GeneReviews

- Cohen Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1482

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Cohen+syndrome%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28cohen+syndrome%5BTIAB%5D%29+OR+%28pepper+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- COHEN SYNDROME
  http://omim.org/entry/216550

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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/cohen-syndrome

Reviewed: January 2008
Published: March 21, 2017

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